SZABO SCANDIC

Part of Europa Biosite

Produktinformation

- Forschungsprodukte & Biochemikalien
- Zellkultur & Verbrauchsmaterial
- Diagnostik & molekulare Diagnostik
- Laborgeräte & Service

Weitere Information auf den folgenden Seiten!
See the following pages for more information!

Lieferung & Zahlungsart

siehe unsere Liefer- und Versandbedingungen

Zuschläge

- Mindermengenzuschlag
- Trockeneiszuschlag
- Gefahrgutzuschlag
- Expressversand

SZABO-SCANDIC HandelsgmbH

Quellenstraße 110, A-1100 Wien

T. +43(0)1 489 3961-0

F. +43(0)1 489 3961-7

mail@szabo-scandic.com

www.szabo-scandic.com

linkedin.com/company/szaboscandic

SANTA CRUZ BIOTECHNOLOGY, INC.

SLC48A1 (h): 293T Lysate: sc-117109

SANTA CRUZ BIOTECHNOLOGY

The Power to Question

BACKGROUND

SLC48A1 is a 239 amino acid protein that is encoded by a gene located on chromosome 12. Encoding over 1,100 genes within 132 million bases, chromosome 12 makes up about 4.5% of the human genome. A number of skeletal deformities are linked to chromosome 12 including hypochondrogenesis, achondrogenesis and Kniest dysplasia. Noonan syndrome, which includes heart and facial developmental defects among the primary symptoms, is caused by a mutant form of PTPN11 gene product, SH-PTP2. Chromosome 12 is also home to a homeobox gene cluster which encodes crucial transcription factors for morphogenesis, and the natural killer complex gene cluster encoding C-type lectin proteins which mediate the NK cell response to MHC I interaction. Trisomy 12p leads to facial development defects, seizure disorders and a host of other symptoms varying in severity depending on the extent of mosaicism and is most severe in cases of complete trisomy.

REFERENCES

1. Allen, T.L., et al. 1996. Cytogenetic and molecular analysis in trisomy 12p. Am. J. Med. Genet. 63: 250-256.
2. Yang, W. and Cole, W.G. 1998. Low basal transcripts of the COL2A1 collagen gene from lymphoblasts show alternative splicing of exon 12 in the Kniest form of spondyloepiphyseal dysplasia. Hum. Mutat. 1: S1-S2.
3. Trowsdale, J., et al. 2001. The genomic context of natural killer receptor extended gene families. Immunol. Rev. 181: 20-38.
4. Zumkeller, W., et al. 2004. Genotype/phenotype analysis in a patient with pure and complete trisomy 12p. Am. J. Med. Genet. A 129: 261-264.
5. Kelley, J., et al. 2005. Comparative genomics of natural killer cell receptor gene clusters. PLoS Genet. 1: e27.
6. Nishimura, G., et al. 2005. The phenotypic spectrum of COL2A1 mutations. Hum. Mutat. 26: 36-43.
7. Segel, R., et al. 2006. The natural history of trisomy 12p. Am. J. Med. Genet. A 140: 695-703.
8. Stein, R. 2007. Genetics of Noonan syndrome—a new gene, and the search is still on. Clin. Genet. 72: 402-404.
9. van der Burgt, I. 2007. Noonan syndrome. Orphanet J. Rare Dis. 2: 4.

CHROMOSOMAL LOCATION

Genetic locus: SLC48A1 (human) mapping to 12q13.11.

PRODUCT

SLC48A1 (h): 293T Lysate represents a lysate of human SLC48A1 transfected 293T cells and is provided as 100 µg protein in 200 µl SDS-PAGE buffer.

STORAGE

Store at -20° C. Repeated freezing and thawing should be minimized. Sample vial should be boiled once prior to use. Non-hazardous. No MSDS required.

RESEARCH USE

For research use only, not for use in diagnostic procedures.

APPLICATIONS

SLC48A1 (h): 293T Lysate is suitable as a Western Blotting positive control for human reactive SLC48A1 antibodies. Recommended use: 10-20 µl per lane.

Control 293T Lysate: sc-117752 is available as a Western Blotting negative control lysate derived from non-transfected 293T cells.

PROTOCOLS

See our web site at www.scbt.com for detailed protocols and support products.